Molecular cytogenetic characterization of mosaicism for a small supernumerary marker chromosome derived from chromosome 8 or r(8)::p11.22→q11.21:: in an 18-year-old female with short stature, obesity, attention deficit hyperactivity disorder, and intellectual disability

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Abstract

Objective
We present molecular cytogenetic characterization of mosaicism for a small supernumerary marker chromosome (sSMC) derived from chromosome 8.

Materials and Methods
An 18-year-old female presented with short stature, obesity, developmental delay, speech delay, dyslexia, attention deficit hyperactivity disorder, and intellectual disability. Cytogenetic analysis of the peripheral blood revealed a karyotype of 47,XX,+mar[22]/46,XX[18]. Array comparative genomic hybridization and metaphase fluorescence in situ hybridization analyses were performed on the peripheral blood to determine the origin and mosaicism of the sSMC, and quantitative fluorescent polymerase chain reaction was used to exclude uniparental disomy.

Results
Array comparative genomic hybridization analysis of the blood revealed a result of arr 8p11.22q11.21 (39,136,065-49,725,726)×2.80 (Log2 ratio = 0.49), consistent with 70–80% mosaicism, encompassing 33 OMIM genes including GOLGA7, AGPAT6, NKX6-3, KAT6A, and FNTA. The sSMC(8) was r(8)::p11.22→q11.21::. Metaphase fluorescence in situ hybridization analysis using the probes of RP11-754D24 (8p11.21) and RP11-769N21 (8q11.21) showed the sSMC(8) in 12/27 of cultured lymphocytes. Quantitative fluorescent polymerase chain reaction analysis excluded uniparental disomy 8.

Conclusion
Mosaic sSMC(8) derived from r(8)::p11.22→q11.21:: can be associated with obesity, intellectual disability, and attention deficit hyperactivity disorder.